1: NM_000485. Reports Homo sapiens aden...[gi:71773149]

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Comment Features Sequence

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LOCUS       NM_000485                807 bp    mRNA    linear   PRI 22-JUN-2008
DEFINITION  Homo sapiens adenine phosphoribosyltransferase (APRT), transcript
            variant 1, mRNA.
ACCESSION   NM_000485
VERSION     NM_000485.2  GI:71773149
KEYWORDS    .
SOURCE      Homo sapiens (human)
  ORGANISM  Homo sapiens
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            Mammalia; Eutheria; Euarchontoglires; Primates; Haplorrhini;
            Catarrhini; Hominidae; Homo.
REFERENCE   1  (bases 1 to 807)
  AUTHORS   Silva,C.H., Silva,M., Iulek,J. and Thiemann,O.H.
  TITLE     Structural complexes of human adenine phosphoribosyltransferase
            reveal novel features of the APRT catalytic mechanism
  JOURNAL   J. Biomol. Struct. Dyn. 25 (6), 589-597 (2008)
   PUBMED   18399692
  REMARK    GeneRIF: Data indicates that the flexible loop structure adopts an
            open conformation before and after binding of both substrates
            adenine and phosphoribosyl pyrophosphate.
REFERENCE   2  (bases 1 to 807)
  AUTHORS   Di Pietro,V., Perruzza,I., Amorini,A.M., Balducci,A.,
            Ceccarelli,L., Lazzarino,G., Barsotti,P., Giardina,B. and
            Tavazzi,B.
  TITLE     Clinical, biochemical and molecular diagnosis of a compound
            homozygote for the 254 bp deletion-8 bp insertion of the APRT gene
            suffering from severe renal failure
  JOURNAL   Clin. Biochem. 40 (1-2), 73-80 (2007)
   PUBMED   17126311
  REMARK    GeneRIF: APRT assay in a sample of patient hemolysate showed no
            detectable activity of the enzyme (25.56+/-9.55 U/L red blood cells
            in control healthy subjects).
REFERENCE   3  (bases 1 to 807)
  AUTHORS   Ewing,R.M., Chu,P., Elisma,F., Li,H., Taylor,P., Climie,S.,
            McBroom-Cerajewski,L., Robinson,M.D., O'Connor,L., Li,M.,
            Taylor,R., Dharsee,M., Ho,Y., Heilbut,A., Moore,L., Zhang,S.,
            Ornatsky,O., Bukhman,Y.V., Ethier,M., Sheng,Y., Vasilescu,J.,
            Abu-Farha,M., Lambert,J.P., Duewel,H.S., Stewart,I.I., Kuehl,B.,
            Hogue,K., Colwill,K., Gladwish,K., Muskat,B., Kinach,R.,
            Adams,S.L., Moran,M.F., Morin,G.B., Topaloglou,T. and Figeys,D.
  TITLE     Large-scale mapping of human protein-protein interactions by mass
            spectrometry
  JOURNAL   Mol. Syst. Biol. 3, 89 (2007)
   PUBMED   17353931
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REFERENCE 4 (bases 1 to 807)
AUTHORS Bruneel,A., Labas,V., Mailloux,A., Sharma,S., Royer,N., Vinh,J., Pernet,P., Vaubourdolle,M. and Baudin,B.
TITLE Proteomics of human umbilical vein endothelial cells applied to etoposide-induced apoptosis
JOURNAL Proteomics 5 (15), 3876-3884 (2005)
PUBMED 16130169
REFERENCE 5 (bases 1 to 807)
AUTHORS Taniguchi,A., Tsuchida,S., Kuno,S., Mita,M., Machida,T., Ioritani,N., Terai,C., Yamanaka,H. and Kamatani,N.
TITLE Identification of two novel mutations in adenine phosphoribosyltransferase gene in patients with 2,8-dihydroxyadenine urolithiasis
JOURNAL Nucleosides Nucleotides Nucleic Acids 23 (8-9), 1141-1145 (2004)
PUBMED 15571218
REMARK GeneRIF: two novel mutations, G133D and V84M, were found in the APRT gene in Japanese patients with APRT deficiency
REFERENCE 6 (bases 1 to 807)
AUTHORS Kamatani,N., Hakoda,M., Otsuka,S., Yoshikawa,H. and Kashiwazaki,S.
TITLE Only three mutations account for almost all defective alleles causing adenine phosphoribosyltransferase deficiency in Japanese patients
JOURNAL J. Clin. Invest. 90 (1), 130-135 (1992)
PUBMED 1353080
REFERENCE 7 (bases 1 to 807)
AUTHORS Chen,J., Sahota,A., Laxdal,T., Scrine,M., Bowman,S., Cui,C., Stambrook,P.J. and Tischfield,J.A.
TITLE Identification of a single missense mutation in the adenine phosphoribosyltransferase (APRT) gene from five Icelandic patients and a British patient
JOURNAL Am. J. Hum. Genet. 49 (6), 1306-1311 (1991)
PUBMED 1746557
REFERENCE 8 (bases 1 to 807)
AUTHORS Ludwig,H., Kuzmits,R., Pietschmann,H. and Muller,M.M.
TITLE Enzymes of the purine interconversion system in chronic lymphatic leukemia: decreased purine nucleoside phosphorylase and adenosine deaminase activity
JOURNAL Blut 39 (5), 309-315 (1979)
PUBMED 116697
REFERENCE 9 (bases 1 to 807)
AUTHORS Holden,J.A., Meredith,G.S. and Kelley,W.N.
TITLE Human adenine phosphoribosyltransferase. Affinity purification, subunit structure, amino acid composition, and peptide mapping
JOURNAL J. Biol. Chem. 254 (15), 6951-6955 (1979)
PUBMED 457664
REFERENCE 10 (bases 1 to 807)
AUTHORS Johnson,L.A., Gordon,R.B. and Emmerson,B.T.
TITLE Adenine phosphoribosyltransferase: a simple spectrophotometric assay and the incidence of mutation in the normal population
JOURNAL Biochem. Genet. 15 (3-4), 265-272 (1977)
PUBMED 869896
COMMENT REVIEWED REFSEQ: This record has been curated by NCBI staff. The reference sequence was derived from BM423481.1 and BU507629.1.
On Aug 3, 2005 this sequence version replaced gi:4502170.

Summary: Adenine phosphoribosyltransferase belongs to the purine/pyrimidine phosphoribosyltransferase family. A conserved feature of this gene is the distribution of CpG dinucleotides. This enzyme catalyzes the formation of AMP and inorganic pyrophosphate from adenine and 5-phosphoribosyl-1-pyrophosphate (PRPP). It also

produces adenine as a by-product of the polyamine biosynthesis pathway. A homozygous deficiency in this enzyme causes 2,8-dihydroxyadenine urolithiasis. Two transcript variants encoding different isoforms have been found for this gene.

Transcript Variant: This variant (1) represents the longer transcript and encodes the longer isoform (a).

Publication Note: This RefSeq record includes a subset of the publications that are available for this gene. Please see the Entrez Gene record to access additional publications.

COMPLETENESS: complete on the 3' end.

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| | 714-807 | BU507629.1 | 115-208 | |

FEATURES Location/Qualifiers

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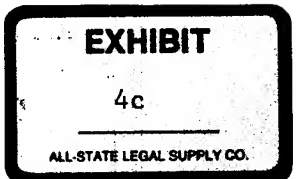

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Last update: Thu, 03 Jul 2008 Rev. 132917